



UMOD gene

uromodulin

Normal Function

The *UMOD* gene provides instructions for making a protein called uromodulin. This protein is produced by the kidneys and then excreted from the body in urine. The function of uromodulin remains unclear, although it is known to be the most abundant protein in the urine of healthy individuals. Researchers have suggested that uromodulin may protect against urinary tract infections. It may also help control the amount of water in urine.

Health Conditions Related to Genetic Changes

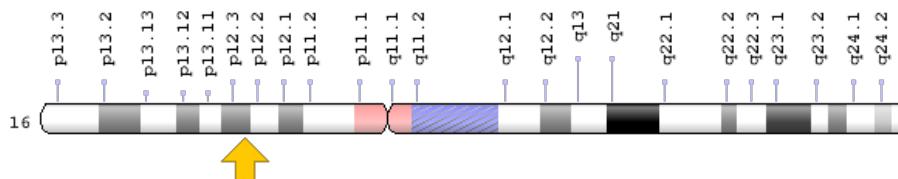
uromodulin-associated kidney disease

More than 40 mutations in the *UMOD* gene have been found to cause uromodulin-associated kidney disease. Most of these mutations change single protein building blocks (amino acids) used to make uromodulin. These mutations alter the structure of the protein, preventing its release from kidney cells. Abnormal buildup of uromodulin may trigger the self-destruction (apoptosis) of cells in the kidneys, causing kidney disease.

Chromosomal Location

Cytogenetic Location: 16p12.3, which is the short (p) arm of chromosome 16 at position 12.3

Molecular Location: base pairs 20,333,051 to 20,356,301 on chromosome 16 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- Tamm-Horsfall protein
- UROM_HUMAN

Additional Information & Resources

GeneReviews

- Autosomal Dominant Tubulointerstitial Kidney Disease, UMOD-Related
<https://www.ncbi.nlm.nih.gov/books/NBK1356>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28UMOD%5BTIAB%5D%29+OR+%28uromodulin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- UROMODULIN
<http://omim.org/entry/191845>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_UMOD.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=UMOD%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=12559
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/7369>
- UniProt
<http://www.uniprot.org/uniprot/P07911>

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